REMARKS

Claims 1-11, 21 and 23-53 are pending before the Office.

I. Non-Compliance to Correction of Sequence Listing Requirement

Examiner has noted the incorrect version of the corrected sequence listing, filed in response to the Paper mailed December 26, 2007. Applicant has cured such defect by way of amendment. Specifically, Applicant timely submitted a preliminary amendment on September 19, 2008 to comply with this requirement. Accordingly, Applicant respectfully submits the earlier requirement for compliance has been met.

II. Objection to the Drawings

Examiner has objected to several drawings for failing to show several features in color. Applicant has cured such defect by way of amendment. Specifically, Figure 2 is now submitted in color. Examiner notes that Figure 1 appears in color. However, there is only black and grey shading within the Figure, with no other color present. This is made clearer in replacement Figure 1, submitted concurrently with this response by way of the aforementioned amendment. Additionally, Applicant also correctly identifies the sequence identifiers within Figures 4 and 6 through the respective replacement sheets. Accordingly, Applicant respectfully requests Examiner withdraw the objection to the Drawings.

III. Claim Objections

Claim 1 is objected to because of a spelling informality. Specifically, the term "Shwachman" is misspelled. Applicant has cured such defect by way of amendment and requests Examiner to withdraw the noted objection.

IV. Rejection under 35 USC §112

Claims 6 and 53 are rejected under 35 USC §112, 2nd paragraph for alleged indefiniteness. Specifically, the Office alleges the numbering of the subject mutations fail to match the respective positions within the relevant sequence identifiers. Examiner cites, as examples, the mutations 24C>A and 258+2T>C not appearing to occur at the respective positions within SEQ ID NO: 1.

Applicant respectfully directs Examiner's attention to den Dunnen et al., which is a publication describing mutation nomenclature. The publication describes the standards within the art of mutation identification. Specifically, when numbering nucleotides for mutation identification, it is standard practice to begin the numbering of a sequence at the "A" of the ATG-translation initiation codon, making this nucleotide "+1". This numbering is shown, for example, in Figure 5 of the present application and is described in the den Dunnen publication at page 122, col. 1, bullet point entitled "Nucleotide Numbering". Thus, 24C>A does match its proper position in SEQ ID NO. 1. With respect to the mutation 258+2T>C, Applicant refers to page 122, col. 2, paragraph 2, bullet points entitled "intronic nucleotides" and "Substitutions"

of den Dunnen et al. The "258+2T>C" mutation refers to position 258 as the last nucleotide of the exon, with the mutation occurring at position 2 of the following intron, wherein a T is substituted with a C. Therefore, this mutation will not be found in SEQ ID NO. 1, since it does not include the intronic sequences. This mutation is, however, shown in Figure 2, in particular the second sequence shown in Figure 2b. Applicant has submitted the den Dunnen reference concurrently with this response and respectfully requests Examiner withdraw the present rejection under 35 USC §112 as applied to claims 6 and 53.

Claims 1-11, 21 and 53 are rejected under 35 USC §112, 1st paragraph for an alleged lack of enablement for diagnosing SDS based on all SBDS mutations. Specifically, the rejection is based on the notion that the specification lacks enablement for any mutation in SBDS other than 183_184TA>CT, 183_184TA>CT + 258+2T>C, and 258+2T>C.

Applicant respectfully submits that the present claims are not directed to diagnosing SDS based on "all SBDS mutations" as the Examiner alleges, but only SBDS mutations "associated with SDS". The present specification and ensuing data teach diagnosing of any SBDS mutation associated with SDS. The Examiner also alleges that the "specification has not provided definitive primers that can be used for amplification and detection of SBDS mutations since the primers described in the specification do not match those of the sequence listing". Applicant respectfully submits this error has been corrected in light of the preliminary amendment, which was filed subsequent to

the mail date of the present office action. Applicant believes Examiner's allegation was a consequence of the original error in the sequence listing. Specifically, in the original sequence listing, SEQ ID NO. 2 was repeated as SEQ ID NO. 3. Thus, all of the remaining SEQ ID NOs. were misnumbered by one numeral. The specification does provide definitive primers that can be used for amplification and detection of SBDS mutations. Applicant respectfully requests Examiner to review the amended sequence listing and reconsider the outstanding rejection under 35 USC §112.

Applicant respectfully requests Examiner withdraw the present rejections and allow the present claims. Applicant is filing this response concurrently with a petition for a two month extension of time. Examiner is encouraged to contact the undersigned with any questions.

Respectfully submitted,

Date: November 19, 2008

David M. Kohn
Reg. No. 53,150

CATALYST LAW GROUP, APC
9710 Scranton Rd., Suite 170
San Diego, CA 92121
(858) 450-0099
(858) 450-9834 (Fax)

In the Drawings:

Please amend the Drawings as follows:

Replace originally filed Figures 1-4 and 6 with replacement sheets containing the same Figures. The replacement sheets are identical in subject matter to original Figures 1-4 and 6, except that the replacement sheets are of a much higher resolution and contain references to the appropriate sequence identifiers. No new matter is added by way of this amendment.